

İ.Ü. DETAE Genetik Anabilim Dalı Faaliyet Raporu

Uluslararası Yayınlar

1. Upregulation of T-Cell-Specific Transcription Factor Expression in Pediatric T-Cell Acute Lymphoblastic Leukemia (T-ALL). Sayitoğlu M, Erbilgin Y, Hatırnaz Ng O, Yıldız I, Celkan T, Anak S, Devocioğlu O, Aydoğan G, Karaman S, Sarper N, Timur C, Ure U, **Ozbek U**.

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2. Identification of interconnected markers for T-cell acute lymphoblastic leukemia. Maiorov EG, Keskin O, Ng OH, **Ozbek U**, Gursoy A. Biomed Res Int. 2013;2013:210253. doi: 10.1155/2013/210253. Epub 2013 Jul 15.

Impact F: 2.88

3. Expression analysis and clinical correlation of aquaporin 1 and 4 genes in human hippocampal sclerosis. Bebek N, Özdemir Ö, Sayitoglu M, Hatırnaz O, Baykan B, Gürses C, Sencer A, Karasu A, Tüzün E, Üzün I, Akat S, Cine N, Sargin Kurt G, Imer M, **Ozbek U**, Canbolat A, Gökyigit A. J Clin Neurosci. 2013 Nov;20(11):1564-70. doi: 10.1016/j.jocn.2012.12.023. Epub 2013 Aug 6.

Impact F: 1.306

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Impact F: 1.013

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Impact F: 0.176

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Impact F: 2.286

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Impact F: 7.251

8. Familial acromegaly due to aryl hydrocarbon receptor-interacting protein (AIP) gene mutation in a Turkish cohort. Mutlu Niyazoglu, Muge Sayitoglu, Sinem Firtina, Esra Hatipoglu, Nurperi Gazioglu, Pinar Kadioglu, Pituitary, 2013, DOI 10.1007/s11102-013-0493-1.

(impact factor 2,66)

9. Renin-Angiotensin System (RAS) Expressions in Myeloid Leukemic Cell Lines. Burak UZ, Suzin C. TATONYAN, Muge SAYITOGLU, Yucel ERBILGIN, Ozden HATIRNAZ NG, Yahya BUYUKASIK, Salih AKSU, Nilgun SAYINALP, Hakan GOKER, Osman I. OZCEBE, Ugur OZBEK, Ibrahim C. HAZNEDAROGLU· International Journal of Hematology and Oncology, Number: 4 Volume: 23 Year: 2013

Impact Factor:

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(impact F: 2.464)

11. Kazancioglu HO, Ak G, Turkmen A, Ozbek U, Tuncer FN, Karabulut A., The Role of MDR1 C3435T Gene Polymorphism on Gingival Hyperplasia in Turkish Renal Transplant Patients Treated With Cyclosporine in the Absence of Calcium Channel Blockers, Transplant Proc. 2013 Jun 8. doi:pii: S0041-1345(12)01334-6. 10.1016/j.transproceed.2012.12.007.

(Impact F: 1.013)

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Impact Factor, 2012: 3.485

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Impact Factor, 2012: 4.162

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Impact Factor:2012: 3.033

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Impact Factor: 0.75

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Impact Factor: 51.658

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Impact Factor:2.557

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Impact Factor: 6.166

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Impact factor: 1.216

21. The relationship between endothelial nitric oxide synthase 4a/4b gene polymorphism and premature coronary artery disease. Ekmekçi A, Ozcan KS, Güngör B, Abaci N, Osmonov D, Zencirci A, Toprak E, Dönmez C, **Ustek D**, Güleç C, Eren M. *Acta Cardiol.* 2013 Oct;68(5):464-8.

Impact factor: 0.607

22. MEFV gene variations in patients with systemic lupus erythematosus. Erer B, Cosan F, Oku B, **Ustek D**, Inanc M, Aral O, Gul A. *Mod Rheumatol.* 2014 Jan;24(1):93-6. doi: 10.3109/14397595.2013.854049.

Impact factor: 1.716

23. Role of the line probe assay INNO-LiPA HBV DR and ultradeep pyrosequencing in detecting resistance mutations to nucleoside/nucleotide analogues in viral samples isolated from chronic hepatitis B patients. Mese S, Arikan M, Cakiris A, Abaci N, Gumus E, Kursun O, Onel D, **Ustek D**, Kaymakoglu S, Badur S, Yenen OS, Bozkaya E. *J Gen Virol.* 2013 Dec;94(Pt 12):2729-38. doi: 10.1099/vir.0.053041-0. Epub 2013 Sep 17.

Impact factor: 3.127

24. Effects of curcumin on proinflammatory cytokines and tissue injury in the early and late phases of experimental acute pancreatitis. Gulcubuk A, Haktanir D, Cakiris A, **Ustek D**, Guzel O, Erturk M, Karabagli M, Akyazi I, Cicekci H, Altunatmaz K, Uzun H, Ates K. *Pancreatol.* 2013 Jul-Aug;13(4):347-54. doi: 10.1016/j.pan.2013.05.005. Epub 2013 May 21.

Impact factor: 2.275

25. Pendrin expression in nodular and non-nodular thyroid tissues. Temel B, Gul N, Kutluturk F, Kapran Y, Sirma S, **Ustek D**, Tunca F, Giles Y, Alagol F. *Endokrynol Pol.* 2013;64(3):208-14.

Impact factor: 1.07

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Aras Y, Sencer A, Gokyigit A, Bilgic B, **Ustek D**. *Gene*. 2013 Oct 15;529(1):190-4. doi: 10.1016/j.gene.2013.06.077. Epub 2013 Jul 18.

Impact factor: 2.196

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Impact factor: 1.107

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Impact factor: 0.948

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Impact factor: 9.737

30. Genome-wide association analysis identifies new susceptibility loci for Behçet's disease and epistasis between HLA-B*51 and ERAP1. Kirino Y, Bertsias G, Ishigatsubo Y, Mizuki N, Tugal-Tutkun I, Seyahi E, Ozyazgan Y, Sacli FS, Erer B, Inoko H, Emrence Z, Cakar A, Abaci N, **Ustek D**, Satorius C, Ueda A, Takeno M, Kim Y, Wood GM, Ombrello MJ, Meguro A, Gül A, Remmers EF, Kastner DL. *Nat Genet*. 2013 Feb;45(2):202-7. doi: 10.1038/ng.2520. Epub 2013 Jan 6.

Impact factor: 35.209

31. The relationship between coronary artery ectasia and eNOS intron 4a/b gene polymorphisms. Ekmekçi A, Ozcan KS, Abaci N, Güngör B, Osmonov D, Tosu R, Toprak E, Güleç C, **Ustek D**, Oz D, Eren M. *Acta Cardiol*. 2013 Feb;68(1):19-22.

Impact factor: 0.607

32. Characterization of H3K9me3- and H4K20me3-associated circulating nucleosomal DNA by high-throughput sequencing in colorectal cancer. Gezer U, **Ustek D**, Yörüker EE, Cakiris A, Abaci N, Leszinski G, Dalay N, Holdenrieder S. *Tumour Biol*. 2013 Feb;34(1):329-36. doi: 10.1007/s13277-012-0554-5. Epub 2012 Oct 20.

Impact factor: 2.518

33. Draft genome sequence of *Halomonas smyrnensis* AAD6T. Sogutcu E, Emrence Z, Arikan M, Cakiris A, Abaci N, Öner ET, **Ustek D**, Arga KY. *J Bacteriol.* 2012 Oct;194(20):5690-1.

Impact factor: 3.194

34. Association of the MEFV gene variations with inflammatory bowel disease in Turkey. Akyuz F, Besisik F, **Ustek D**, Ekmekçi C, Uyar A, Pinarbasi B, Demir K, Ozdil S, Kaymakoglu S, Boztas G, Mungan Z, Gul A. *J Clin Gastroenterol.* 2013 Mar;47(3):e23-7. doi: 10.1097/MCG.0b013e3182597992.

Impact factor: 3.203

35. TT polymorphism in rs2165241 and rs1048661 region in lysyl oxidase like-1 gene may have a role in stress urinary incontinence physiopathology. Ozbek E, Polat EC, Ozcan L, Otunctemur A, Emrence Z, **Ustek D**. *J Obstet Gynaecol Res.* 2013 Jan;39(1):237-42. doi: 10.1111/j.1447-0756.2012.01942.x. Epub 2012 Jul 6.

Impact factor: 0.546

36. The association of TNFRSF1A gene and MEFV gene mutations with adult onset Still's disease. Cosan F, Emrence Z, Erbag G, Azakli H, Yilmazer B, Yazici A, Ekmekci SS, Abaci N, **Ustek D**, Cefle A. *Rheumatol Int.* 2013 Jul;33(7):1675-80. doi: 10.1007/s00296-012-2609-8. Epub 2012 Dec 27.

Impact factor: 2.214

Uluslararası sözü sunumlar

1. Next-Generation Sequencing Of The BCR-ABL1 Kinase Domain May Be Beneficial In Decision Making Among Chronic Myeloid Leukemia Patients With Tyrosine Kinase Inhibitor Resistance. Yücel Erbilgin, Ahmet Emre Eşkazan, Özden Hatırnaz Ng, Ayşe Salihoğlu, Tuğrul Elverdi, Sinem Fırtına, Suzin Çatal Tatonyan, Şeniz Öngören Aydın, Muhlis Cem Ar, Zafer Başlar, Müge Sayitoğlu, Burhan Ferhanoğlu, Yıldız Aydın, Uğur Özbek, Teoman Soysal. ASH Meeting 7-10 December 2013, New Orleans, USA.
2. Normalization Methods and Analysis Packages for 450k Data Analysis. Yücel Erbilgin. High-Throughput Methylome Analysis & Its Impact on Human Health and Disease. 31 May 2013, Istanbul, Turkey.
3. The Interlaboratory Robustness Of Next-Generation Sequencing (IRON) Study Phase II: Deep-Sequencing Analyses Of Hematological Malignancies

Performed In 8,867 Cases By An International Network Involving 27 Laboratories. Alexander Kohlmann, Giovanni Martinelli, Mary Alikian^{3*}, Valentina Artusi, Bernd Auber, Monika Belickova, Ilaria Bronzini, Giovanni Cazzaniga, Sabina Chiaretti, Thomas Ernst, Marc Fuellgrabe, Christian Gabriel, Jesus Hernandez, Joop H. Jansen, Ilaria Iacobucci, Thomas Lion, Laurence Lode, Joaquin Martinez-Lopez, Ken I. Mills, Maximilian Mossner, Katerina Machova Polakova, Naomi A. Porret, Sarka Pospisilova, Claude Preudhomme, Muge Sayitoglu, Simona Soverini, Orietta Spinelli, Christian Thiede, Peter Vandenberghe, Juh Eng Yeoh, Andreas Hochhaus, and Torsten Haferlach. Type: Oral Session: 611. Leukemias: Biology, Cytogenetics and Molecular Markers in Diagnosis and Prognosis I, 55.th ASH meeting 2013, USA, New Orleans.

4. Working Package ALL: Data from Istanbul. Müge Sayitoglu. Iron II Investigator Meeting, July 10-11 2013, Munich, Germany.
5. Working Package CML: Data from Istanbul. Yücel Erbilgin Iron II Investigator Meeting, July 10-11 2013, Munich, Germany.
6. “Clinical Application on Exome Sequencing Analysis”, Feyza Nur Tuncer, Next Generation Sequencing Hybrid Course, May17-20, 2013, Istanbul, Turkey.
7. “Infinium HD Assay: Application”, Feyza Nur Tuncer High-Throughput Methylome Analysis & Its Impact on Human Health and Disease. 31 May 2013, Istanbul, Turkey.
8. High Throughput Methylome Analysis & Its Impact on Human Health and Disease., ‘*IntenC Project: Genome-wide integrated methylome analysis in idiopathic generalized epilepsies*’, Uğur Özbek, Taksim Peak Hotel, 31.05.2013, İstanbul
9. Haytural H, Yalcinkaya N, Ulusoy C, Kucukerden M, Turan S, Sehitoglu E, Ugurel E, Cavus F, Vural B, Tüzün E. MTCH1 antibodies in neuro-Behcet’s disease (Oral presentation, English). 4th International Medical Students’ Research Congress (2013, Istanbul, Turkey) Abstract Booklet; p:46
10. Haytural H, Yalcinkaya N, Ulusoy C, Kucukerden M, Turan S, Sehitoglu E, Ugurel E, Cavus F, Vural B, Tüzün E. MTCH1 antibodies in neuro-Behcet’s disease (Oral presentation, English). 4th International Medical Students’ Research Congress (2013, Istanbul, Turkey) Abstract Booklet; p:46.

Uluslararası sunulan posterler

1. **N. Coban,** C. Gulec, B. Ozsait-Selcuk, N. Erginel-Unaltuna. Regulation of Expression of CYP19A1, MIF, ABCA1 by the Retinoic Acid Receptor-Related Orphan Receptor α (ROR α). European Human Genetics Conference 2013, June 8-11 2013, Paris. European Journal of Human Genetics 2013;21 (suppl 2):131, P04.03.
2. **N. Erginel-Unaltuna,** C. Gulec, N. Coban, B. Ozsait, S. Sirma-Ekmekci. ROR-Alpha Target Genes in Monocyte and Endothelium. European Human Genetics Conference 2013, June 8-11 2013, Paris. European Journal of Human Genetics 2013;21 (suppl 2):460, P20.48.
3. **C. Gulec,** N. Coban, B. Ozsait, S. Sirma-Ekmekci, N. Erginel-Unaltuna. Inhibitory Effect of SR1001 on ROR-Alpha Activity Requires Intracellular Cholesterol. European Human Genetics Conference 2013, June 8-11 2013, Paris. European Journal of Human Genetics 2013;21 (suppl 2):461, P20.49.
4. **F. Guclu** - Geyik, T. Erginel, N. Erginel- Unaltuna. ESR1 gene promoter methylation in adipose tissue, European Human Genetics Conference 2013, Paris, June 8-11 2013, European Journal of Human Genetics 2013;21 (suppl 2):550, J15.08
5. **B. Ozsait-Selcuk,** B. Yuzbasiogullari, N. Erginel-Unaltuna. Functional analysis of Midnolin and Taube Nuss genes: Potential approach for identification of cardiac specific physiological processes. European Society of Human Genetics, (ESHG), Paris, 8-11 June 2013, European Journal of Human Genetics 2013;21 (suppl 2):**604, J20.11**
6. **E. Komurcu Bayrak,** M. Poda, A. Onat, V. Sansoy, B. Yuzbasiogullari, T. Lehtimaki, S. Humphries, N. Erginel Unaltuna. Gene-gene interaction between APOE and USF1 for coronary heart disease in TARF study.

European Society of Human Genetics, (ESHG), Paris, 8-11 June 2013,
European Journal of Human Genetics 2013;21 (suppl 2):604, P16.020

7. Genetic and Epigenetic Profile Of Early Relapsed Childhood ALL
Yucel Erbilgin, Ozden Hatirnaz Ng, Sinem Firtina, Tiraje Celkan, Sema Anak, MD, Zeynep Karakas, Aykan Ozguven, MD4*, Nazan Sarper, MD5*, Emine Zengin, Emine Turkkan, Didem Yalcin Atay, MD6*, Yuk Yin Ng, PhD1,7*, Omer Dogru, Muge Sayitoglu, and Ugur Ozbek. ASH Meeting 7-10 December 2013, New Orleans, USA. Blood October 21, 2013 vol. 122 no. 21 1380
8. Simona Soverini, Thomas Ernst, Alexander Kohlmann, Caterina De Benedittis, Mary Alikian, Letizia Foroni, Katerina Machova Polakova, Adela Brouckova, Yucel Erbilgin, Ozden Hatirnaz, Ugur Ozbek, Cristina Robledo, Jesus Maria Hernandez, Milena Musilova, Santiago Barrio, Joaquin Martinez-Lopez, Renate Kastner, Thomas Lion, Torsten Haferlach, Andreas Hochhaus, and Giovanni Martinelli. Sensitivity, Reproducibility and Clinical Utility Of Next-Generation Sequencing (NGS) for BCR-ABL1 Kinase Domain Mutation Screening: Results From The CML Work Package Of The Iron-II (Interlaboratory ROBustness Of Next-Generation Sequencing) International Study. Blood 2013 122:3824. ASH Meeting 7-10 December 2013, New Orleans, USA.
9. Next-Generation Sequencing of the BCR-ABL1 Kinase Domain in Resistant CML Patients. Yucel Erbilgin, Ozden Hatirnaz Ng, Seniz Ongoren Aydin, Sinem Firtina, Ismail Can, Teoman Soysal, Muge Sayitoglu, Ugur Ozbek. Otto Warburg International Summer School and Research Symposium 2013 Next Generation Sequencing and its Impact on Genetics. August 19th-26th, Berlin, Germany.
10. Whole Genome Methylation Profile of Relapsed Childhood Leukemia. Yücel Erbilgin, Özden Hatirnaz Ng, Sinem Firtina, Tülin Tiraje Celkan, Saniye Sema Anak, Zeynep Karakaş, Aykan Güven, Nazan Sarper, Emine Zengin, Emine Türkkan, Didem Yalçın Atay, Yuk Yin Ng, Ömer Doğru, Türkan Tansel, Enver Dayıoğlu, Müge Sayitoğlu, Uğur Özbek. Translating Epigenomes into Function: a Next-Generation Challenge for Human Disease. October 13-16, 2013, Capri, Italy.

11. Coban A, Cavus F, Ulusoy C, Turan S, Vural B, Turkoglu R, Kurtuncu M, Eraksoy M, Akman-Demir G, Tüzün E. MTCH1 antibodies in neuromyelitis optica patients with or without aquaporin-4 antibodies (Poster presentation, English). 23rd Meeting of ENS (2013, Barcelona, Spain) Abstract Booklet (Journal of Neurology, suppl. 1, supplement to volume 260, 2013); p:S115.
12. F.Yesim Kesim, Feyza N. Tuncer , Emrah Yücesan, Ozkan Ozdemir, Sibel Ugur Iseri, Ugur Ozbek. Trisomy 21 Revealed in a Linkage Scan with Whole Genome SNP Genotyping. ESHG 26th course in Medical Genetics, May 12-16, 2013, Bertinoro di Romagna, Italy.
13. F. Tuncer, S. A. Ugur Iseri, M. Calik, A. O. Caglayan, A. Iscan, M. Gunel, U. Ozbek. Linkage analysis and exome sequencing in a large highly inbred consanguineous kindred to identify idiopathic generalized epilepsy genes. American Society of Human Genetics October 22-26, 2013, Boston, USA.

Ulusal Faaliyetler

Ulusal Yayınlar

Özsait B., Ünaltuna-Erginel N. Farklı spermatogenik düzensizliklere sahip hastaların spermatozoa mikroRNA ekspresyon profillerindeki değişim (Güncel Makale Özeti) Androloji Bülteni 2013: 54:198

Ulusal sözlü sunumlar

1. Tirozin Kinaz İnhibitörlerine Dirençli Kronik Miyeloid Lösemi Hastalarında Yeni Nesil Dizileme Tekniği İle BCR-ABL1 Kinaz Bölgesi Mutasyonlarının Tespiti. Yücel Erbilgin, Ahmet Emre Eşkazan, Özden Hatırnaz Ng, Ayşe Salihoğlu, Tuğrul Elverdi, Sinem Fırtına, Suzin Çatal Tatonyan, Şeniz Öngören Aydın, Muhlis Cem Ar, Zafer Başlar, Müge Sayitoğlu, Burhan Ferhanoglu, Yıldız Aydın, Uğur Özbek, Teoman Soysal. 23-26 Ekim, 39. Ulusal Hematoloji Kongresi, Antalya, Türkiye.

2. Çocukluk Çağı Nüks Akut Lenfoblastik Lösemi Hastalarında Tüm Genom SNP Array Analizi. Yücel Erbilgin, Özden Hatırnaz Ng, Tülin Tiraje Celkan, Sinem Fırtına, İsmail Can, Ayşegül Ünüvar, Leyla Ağaoğlu, Hilmi Apak, Emine Zengin, Emine Türkkan, Müge Sayitoğlu, Uğur Özbek. 9. Ulusal Pediatrik Hematoloji Kongresi 24-28 Mayıs 2013, Van, Türkiye.
3. İstanbul Üniversitesi DETAE Genetik AD Moleküler Hematoloji Çalışma Deneyimleri, Müge Sayitoğlu. 1. Hematolojik Genetik Sempozyumu, 2-4 Aralık 2013 İzmir
4. Alkac M, İşbilen M, Güre AO, Vural B, Üçlü Negatif Meme Kanserin (ÜNMK) tedavisinde AZD7762 ilacının hassaiyetinin belirlenmesinde RBFOX2 geninin marker olarak kullanımının araştırılması, 5. DETAE Günleri, 11-12 Kasım 2013, İstanbul, Türkiye

B. Ulusal sunulan posterler

1. **Neslihan Abacı**, Muzaffer Arıkan, Emre Gök, Aris Çakiris, Sema Sırma Ekmekçi, Türkan Tansel, Duran Üstek. Konjenital kalp defektli hastalarda yeni nesil dizileme yöntemi ile mitokondriyal mutasyonalar. XIII. Ulusal Tıbbi Biyoloji ve Genetik Kongresi. PS-11 17, 27-30 Ekim 2013 , Kuşadası
2. **Sema Sırma Ekmekçi**, Neslihan Abacı, Çağrı Güleç, Zeliha Emrence, Cumhur Gökhan Ekmekçi, Duran Üstek. P53'ün posttranslasyonel regülatörü olan HEP27'nin ekspresyonu LEF1 tarafından kontrol edilmektedir. XIII. Ulusal Tıbbi Biyoloji ve Genetik Kongresi. PS-07 08, 27-30 Ekim 2013 , Kuşadası
3. **Neslihan Çoban**, Altan Onat, Günay Can, Nihan Erginel Ünaltuna. Genetic Variation in LPA Gene Predicts Plasma Lipoprotein(a) Level and Type 2 Diabetes Risk. Journal of the American College of Cardiology, Volume 62, Issue 18, Supplement 2, 29 October 2013, Page C53. 29. Ulusal Kardiyoloji Kongresi, 2013, Antalya
4. **Neslihan Çoban**, Altan Onat, Günay Can, Nihan Erginel Ünaltuna. A Genetic Variant Associated with Lipoprotein(a) Level and Coronary Disease Risk in

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5. **Filiz Geyik**, Neslihan Çoban, Berna Yüzbaşıoğulları, Altan Onat, Vedat Sansoy, Can Günay, Nihan Erginel Ünaltuna. Gene-Gene Interaction between APOA4 and FTO for Obesity in TARF Study. Journal of the American College of Cardiology, Volume 62, Issue 18, Supplement 2, 29 October 2013, Page C53. 29. Ulusal Kardiyoloji Kongresi, 2013, Antalya
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